



PEER-REVIEW REPORT

Name of journal: *World Journal of Clinical Cases*

Manuscript NO: 79958

Title: Novel TINF2 gene mutation in dyskeratosis congenita with extremely short telomeres: A case report

Provenance and peer review: Unsolicited Manuscript; Externally peer reviewed

Peer-review model: Single blind

Reviewer's code: 06139976

Position: Peer Reviewer

Academic degree: Doctor, MD

Professional title: Doctor, Professor

Reviewer's Country/Territory: Japan

Author's Country/Territory: Mexico

Manuscript submission date: 2022-09-13

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-09-14 00:16

Reviewer performed review: 2022-09-15 10:55

Review time: 1 Day and 10 Hours

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input type="checkbox"/> Minor revision <input checked="" type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input checked="" type="checkbox"/> Yes <input type="checkbox"/> No



Peer-reviewer statements	Peer-Review: [<input type="checkbox"/>] Anonymous [<input checked="" type="checkbox"/>] Onymous Conflicts-of-Interest: [<input type="checkbox"/>] Yes [<input checked="" type="checkbox"/>] No
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SPECIFIC COMMENTS TO AUTHORS

This is an excellent case report of dyskeratosis congenita with a novel genetic variant. The case presentation and discussion are generally well described. I would like to make a few recommendations to improve this case report.

1. Dyskeratosis congenita and collagen disease The authors describe dyskeratosis congenita as a rare genetic disease. For readers who do not specialise in this field, the frequency of this disease should also be explained. Collagen diseases such as dermatomyositis and systemic lupus erythematosus also come up in the differential as diseases that generally present with haematopoietic and skin disorders in young females. Have screening tests such as antinuclear antibodies been performed for these diseases?
2. Differentiating fungal infections The patient was treated with high-dose prednisolone. Prednisolone can cause a high risk of infection. Was fungal infection considered as a differential for the white lesions on the skin and tongue in that patient? In Figure 1C, light reflections and leukokeratosis plaques on the surface of the tongue are difficult to distinguish. Also, the oral cavity is dark and difficult to see. Therefore, a photograph in which the palate or posterior wall of the pharynx is illuminated rather than the surface of the tongue is more desirable.
- 3 Genetic counselling It is excellent that an accurate diagnosis was made promptly in this case. As the authors point out, there is no fundamental treatment for this disease. Has appropriate mental care and support been provided for these patients? For example, were regular genetic counselling or consultation to a psychiatrist considered?



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Reviewer's code: 03604107

Position: Editorial Board

Academic degree: MD, PhD

Professional title: Professor

Reviewer's Country/Territory: Albania

Author's Country/Territory: Mexico

Manuscript submission date: 2022-09-13

Reviewer chosen by: AI Technique

Reviewer accepted review: 2022-09-14 04:16

Reviewer performed review: 2022-09-17 11:48

Review time: 3 Days and 7 Hours

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input checked="" type="checkbox"/> Grade B: Very good <input type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input type="checkbox"/> Accept (General priority) <input checked="" type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Re-review	<input type="checkbox"/> Yes <input checked="" type="checkbox"/> No



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Peer-reviewer statements	Peer-Review: <input checked="" type="checkbox"/> Anonymous <input type="checkbox"/> Onymous Conflicts-of-Interest: <input type="checkbox"/> Yes <input checked="" type="checkbox"/> No
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SPECIFIC COMMENTS TO AUTHORS

This is a good paper, on a very interesting topic. Since the condition is rare and genetic, a table with review of most important literature sources would be good for readers. Also, there is no language editing certificate for non-native English authors included in the system. Can you provide one?



RE-REVIEW REPORT OF REVISED MANUSCRIPT

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Peer-review model: Single blind

Reviewer's code: 06139976

Position: Peer Reviewer

Academic degree: Doctor, MD

Professional title: Doctor, Professor

Reviewer's Country/Territory: Japan

Author's Country/Territory: Mexico

Manuscript submission date: 2022-09-13

Reviewer chosen by: Ji-Hong Liu

Reviewer accepted review: 2022-10-13 10:32

Reviewer performed review: 2022-10-13 10:50

Review time: 1 Hour

Scientific quality	<input type="checkbox"/> Grade A: Excellent <input type="checkbox"/> Grade B: Very good <input checked="" type="checkbox"/> Grade C: Good <input type="checkbox"/> Grade D: Fair <input type="checkbox"/> Grade E: Do not publish
Language quality	<input type="checkbox"/> Grade A: Priority publishing <input checked="" type="checkbox"/> Grade B: Minor language polishing <input type="checkbox"/> Grade C: A great deal of language polishing <input type="checkbox"/> Grade D: Rejection
Conclusion	<input type="checkbox"/> Accept (High priority) <input checked="" type="checkbox"/> Accept (General priority) <input type="checkbox"/> Minor revision <input type="checkbox"/> Major revision <input type="checkbox"/> Rejection
Peer-reviewer	Peer-Review: <input type="checkbox"/> Anonymous <input checked="" type="checkbox"/> Onymous



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statements

Conflicts-of-Interest: [] Yes [Y] No

SPECIFIC COMMENTS TO AUTHORS

The authors responded appropriately to the reviewers' comments. The manuscript is considered to meet the criteria for accept.